

Relationship Testing TECHNICAL REPORT FOR TESTING IN 2020



ABSTRACT

AABB surveyed facilities accredited for Relationship Testing (RT) activities for data of interest to the RT community. The total volume of cases tested and reported in 2020 was 337,931. Of those, 46% were legal chain of custody cases for nonimmigration purposes and demonstrated an exclusion rate of 22.45%. Of the total case volume, 4% were for immigration, visa, passport or citizenship cases with an exclusion rate of 4.63%; 50% were unaccredited cases lacking a chain of custody tested for curiosity and showed an exclusion rate of 29.17%. Of all samples collected, more than 97% were buccal swabs. DNA analysis of autosomal short tandem repeats comprised 97% of the tests performed. X- chromosome analysis was performed in addition to the autosomal analyses on 19% of the cases, a small number of cases also received Y Chromosome or mitochondrial analysis. Of the laboratories surveyed, 68.4% incorporate apparent mutations into the combined likelihood ratio by dividing mutation rate by the average probability of exclusion. Twenty-one percent of the laboratories use a method that considers the short tandem repeat differences.

PREFACE

This survey provides information on the state of the RT community, tries to ask questions that may be of interest, and tracks trends in testing. Evaluation of these data was anonymous. AABB scientific staff reviewed the raw data and provided only anonymized aggregate data and tables for review by the Relationship Testing Standards Committee (RTSC) and the Relationship Testing Accreditation Committee (RTAC). Data from AABB-accredited facilities that perform only Collection and Report Verification activities are excluded from this report to avoid any duplication of data submitted by the testing laboratory.

Apparent mutation data submitted for 2020 was not sufficient to provide meaningful mutation frequencies. Many facilities faced significant challenges during the pandemic and several ceased operations for part of 2020. Mutation data will be requested of the labs for the *2021 RT Technical Report.*

ANNUAL VOLUME OF TESTING

The volume reported for cases tested in 2020 was 337,257, compared to 410,931 in 2019. Because some laboratories did not provide data, this is an underestimate of the actual number of cases tested by AABB-accredited laboratories. In addition to volume of accredited tests, laboratories were asked if they tested cases where the chain of custody did not meet the requirements of the *AABB Standards for Relationship Testing*. The tested individuals, without a proper witness (see Standards), generally self-collect these so called "non-legal" tests. AABB has taken the position that it cannot prohibit accredited laboratories from performing these types of tests but reminds laboratories that they cannot claim or advertise that their "non-legal" testing meets AABB standards. This includes reports that state the "testing" meets the standards and only the chain of custody is lacking. Laboratories must conform in all aspects and cannot choose standards to which they will adhere.

Table 1 indicates the volumes of cases reported by case type in comparison to 2019.

TABLE 1. CASES REPORTED BY TYPE

Case Type	Cases Reported 2020	% Total 2020	% Total 2019
Non-Immigration Legal	155,672	46.16%	57.56%
Immigration, Visa, Passport	11,860	3.52%	5.74%
Non-legal / No Chain of Custody	169,726	50.33%	36.70%

LABORATORIES BY SIZE

Table 2 indicates the size of the various responding laboratories by volume of cases reported.

TABLE 2. LABORATORY SIZE BY THE VOLUME OF CASES REPORTED

Number of Cases Reported	Percentage of RT Laboratories		
	2020	2019	
<100	38.89%	19.05%	
100-1,000	16.67%	33.33%	
1,001-10,000	22.22%	33.33%	
10,001-100,000	11.11%	4.76%	
>100,000	11.11%	9.52%	

EXCLUSION RATE

We asked laboratories to report exclusions, or hypotheses not supported for non- parentage cases, separately by case type. The observed rate of exclusion varies significantly depending on the type of case as shown in Table 3.

TABLE 3. EXCLUSIONS REPORTED BY CASE TYPE

Exclusions(or hypothesis not supported)	Non-Immigration Legal	Immigration, Visa, Passport	Non-legal / No Chain of Custody
Average Exclusion Rate	22.45%	4.63%	29.17%
Median Exclusion Rate	21.76%	2.75%	25.91%
Standard Deviation	10.38%	3.18%	10.85%
Range	0-27.27%	0-9.8%	0-42.86%

MISCONCEPTIONS IN PATERNITY TESTING – EXCLUSION RATE

AABB has observed misinterpretation of data reported for exclusion rates in previous reports. It is important to clarify what the exclusion rate does not represent. An exclusion rate of 30% does not mean that 30% of fathers are raising children that are not biologically theirs. From the data, it can only be concluded that, of the people who needed a relationship test, some percentage of those tests either exclude or do not support the tested relationship. Additionally, there are many situations in which the relationship was never in question, but a DNA test was necessary to provide proof of relationship for legal reasons.

COMBINED RELATIONSHIP INDEX (COMBINED LIKELIHOOD RATIO)

The laboratories were asked to indicate what combined relationship index (CRI) they considered acceptable for cases with a standard trio (mother, child, father), single parent cases (mother (or father) not tested cases), and family study / reconstruction cases of more than two tested parties (cases where the disputed parent is missing and other relatives are used to evaluate parentage).

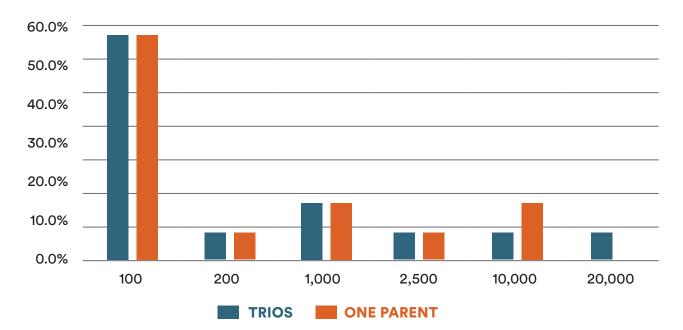
The *AABB Standards for Relationship Testing Laboratories* sets the minimum CRI (W) for parentage cases at 100. An index of 100 is reliable, but indices of higher values can be obtained using current methods. There has been a tendency for laboratories to set much higher values as a minimum likelihood ratio, such as 10,000 to 1 and 100,000 to 1 for some of their tested hypotheses, but not all (such as family study/reconstruction cases). Although setting these higher standards for internal use is not inappropriate, it is inappropriate to claim lower values are not reliable. The minimum acceptable CRI for parentage cases, by policy, is in excess of the AABB standard for 42.9% of the laboratories. One laboratory reporting using a lower CRI for single parent cases than that used for standard trios.

For the 2020 RT Technical Report, data was not collected on minimum CRI for two party comparisons of full siblings, half siblings, avuncular, and single grandparentage likelihood ratios. Beginning with the 13th Edition of the AABB Standards for Relationship Testing Laboratories, minimum CRI standards are defined for two party non parentage comparisons.

TABLE 4A. LABORATORIES' MINIMUM COMBINED LIKELIHOOD RATIOS (% OF LABORATORIES USING A W AS THEIR MINIMUM) FOR STANDARD TRIOS, ONE PARENT (MOTHER OR FATHER NOT TESTED), FAMILY STUDIES >2 PARTIES

W	Trio	One Parent	Family Study >2 parties
whatever is obtained			0.00%
10			78.57%
80			7.14%
100	57.14%	57.14%	7.14%
200	7.14%	7.14%	
1,000	14.29%	14.29%	
2,500	7.14%	7.14%	
10,000	7.14%	14.29%	
20,000	7.14%		

TABLE 4B. LABORATORIES' MINIMUM COMBINED LIKELIHOOD RATIOS(% OF LABORATORIES USING A W AS THEIR MINIMUM)FOR STANDARD TRIOS AND ONE PARENT CASES



TECHNOLOGY USE

Table 5 provides a breakdown of the technology used to resolve the reported cases.

Technology / Method	Utilization
DNA-STR	97.12%
X Chromosome Analysis	19.80%
Non-Invasive Prenatal Paternity	1.88%
Y Chromosome Analysis	0.19%
Mitochondrial Analysis	0.01%
DNA-SNP Array	none

none

TABLE 5. THE TECHNOLOGY USED IN CASES REPORTED IN 2020

SAMPLE SOURCE

DNA-NGS

Laboratories reported approximately 771,573 samples used for casework in 2020. This total includes non-legal cases and samples collected without a chain of custody, any of the following sample types lacking a chain of custody or do not meet the requirements for identification in the Standards for Relationship Testing Laboratories would not be appropriate for an AABB-accredited legal relationship test. Buccal swabs account for ~98% of the samples. Various other samples were also reported (See Table 6).

TABLE 6. SAMPLE SOURCE IN 2020

Sample Type	Percentage
Buccal Swabs	97.870%
Dried Blood Spots	0.752%
Tissues, body fluids, teeth, cell pellets	0.720%
Liquid Blood	0.530%
Fingernails or swabs other than buccal	0.085%
Hair	0.025%
Paraffin Block	0.011%
Amniotic Fluid	0.004%
Bone	0.003%
Chorionic Villi	0.000%
abandoned DNA (toothbrush, etc.)	0.0001%
received DNA extracts	none

MUTATION CALCULATION

Single inconsistencies are routinely seen in the testing of parentage cases. If a laboratory concludes that the inconsistency is a mutation, then the mutation result must be incorporated into the reported results. Laboratories were asked how they calculated the parentage index (PI) for these loci. Most commonly, laboratories use the mutation rate divided by the average probability of exclusion. Some labs reported using more than one method.

TABLE 7. REPORTED MUTATION CALCULATION METHODS FOR 2020

Mutation calculation method	Usage
Mutation rate/average probability of exclusion	66.67%
Using a method that takes into account STR repeat differences (Brenner's Method)	26.67%
Fimmer's Method	6.67%
Standard PI using the mutation rate as the disputed parent's transmission chance	6.67%
Use the mutation rate as the PI	6.67%
Familias	none

If you have questions regarding this report, please contact accreditation@aabb.org.



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